# Robert William Taylor

List of Publications by Year
in descending order



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|---|---|---|---|
| 241 papers | 17,681 citations | 20817 60 h-index | 17592 121 g-index |
| 241 all docs | 241 docs citations | 241 times ranked | 18027 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130. | 5.3 | 17 |
| 2 | The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. Journal of Raman Spectroscopy, 2022, 53, 172-181. | 2.5 | 5 |
| 3 | Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062. | 2.9 | 3 |
| 4 | Identification and characterization of novel <scp>MPC1</scp> gene variants causing mitochondrial pyruvate carrier deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 264-277. | 3.6 | 7 |
| 5 | Mosaic dysfunction of mitophagy in mitochondrial muscle disease. Cell Metabolism, 2022, 34, 197-208.e5. | 16.2 | 35 |
| 6 | Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38. | 8.2 | 85 |
| 7 | Biallelic variants in TAMM41 are associated with low muscle cardiolipin levels, leading to neonatal mitochondrial disease. Human Genetics and Genomics Advances, 2022, 3, 100097. | 1.7 | 3 |
| 8 | Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Analyst, The, 2022, 147, 2533-2540. | 3.5 | 9 |
| 9 | RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, . | 8.2 | 6 |
| 10 | Defining mitochondrial protein functions through deep multiomic profiling. Nature, 2022, 606, 382-388. | 27.8 | 49 |
| 11 | Neuromuscular Junction Abnormalities in Mitochondrial Disease. Neurology: Clinical Practice, 2021, 11, 97-104. | 1.6 | 10 |
| 12 | LONP1 and mtHSP70 cooperate to promote mitochondrial protein folding. Nature Communications, 2021, 12, 265. | 12.8 | 58 |
| 13 | POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135. | 12.8 | 21 |
| 14 | The molecular pathology of pathogenic mitochondrial tRNA variants. FEBS Letters, 2021, 595, 1003-1024. | 2.8 | 29 |
| 15 | The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442. | 4.5 | 33 |
| 16 | Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104. | 2.3 | 1 |
| 17 | Machine learning algorithms reveal the secrets of mitochondrial dynamics. EMBO Molecular Medicine, 2021, 13, e14316. | 6.9 | 6 |
| 18 | Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417. | 27.0 | 84 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | A novel MT-CO2 variant causing cerebellar ataxia and neuropathy: The role of muscle biopsy in diagnosis and defining pathogenicity. Neuromuscular Disorders, 2021, 31, 1186-1193. | 0.6 | 5 |
| 20 | Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584. | 10.2 | 96 |
| 21 | ATG7 safeguards human neural integrity. Autophagy, 2021, 17, 2651-2653. | 9.1 | 7 |
| 22 | PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425. | 2.4 | 8 |
| 23 | The Effect of tRNA[Ser]Sec Isopentenylation on Selenoprotein Expression. International Journal of Molecular Sciences, 2021, 22, 11454. | 4.1 | 8 |
| 24 | Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. American Journal of Human Genetics, 2021, 108, 2195-2204. | 6.2 | 26 |
| 25 | Emerging roles of ATG7 in human health and disease. EMBO Molecular Medicine, 2021, 13, e14824. | 6.9 | 61 |
| 26 | Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397. | 6.9 | 5 |
| 27 | 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880. | 27.0 | 352 |
| 28 | 2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. Nature Communications, 2021, 12, 6997. | 12.8 | 12 |
| 29 | Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50. | 3.6 | 113 |
| 30 | Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. European Journal of Human Genetics, 2020, 28, 373-377. | 2.8 | 20 |
| 31 | Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308. | 3.6 | 43 |
| 32 | The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209. | 2.4 | 14 |
| 33 | Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286. | 2.0 | 7 |
| 34 | Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101. | 6.2 | 39 |
| 35 | Assessment of mitochondrial respiratory chain enzymes in cells and tissues. Methods in Cell Biology, 2020, 155, 121-156. | 1.1 | 32 |
| 36 | A novel de novo ACTA1 variant in a patient with nemaline myopathy and mitochondrial Complex I deficiency. Neuromuscular Disorders, 2020, 30, 159-164. | 0.6 | 7 |

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|---|---|---|---|
| 37 | A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. Neuromuscular Disorders, 2020, 30, 661-668. | 0.6 | 8 |
| 38 | Earlyâ€onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic COQ8A variants, including a large intragenic deletion. JIMD Reports, 2020, 54, 45-53. | 1.5 | 8 |
| 39 | SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. Molecular Genetics and Metabolism Reports, 2020, 25, 100657. | 1.1 | 10 |
| 40 | <scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659. | 6.9 | 44 |
| 41 | Nuclear genetic disorders of mitochondrial DNA gene expression. , 2020, , 375-409. | | 0 |
| 42 | The genetic basis of isolated mitochondrial complex II deficiency. Molecular Genetics and Metabolism, 2020, 131, 53-65. | 1.1 | 22 |
| 43 | Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248. | 8.8 | 48 |
| 44 | Mitochondrial OXPHOS Biogenesis: Co-Regulation of Protein Synthesis, Import, and Assembly Pathways. International Journal of Molecular Sciences, 2020, 21, 3820. | 4.1 | 74 |
| 45 | The m.15043GÂ>ÂA MT-CYB variant is not a pathogenic mtDNA variant. Journal of the Neurological Sciences, 2020, 417, 116950. | 0.6 | 1 |
| 46 | Novel MT-ND Gene Variants Causing Adult-Onset Mitochondrial Disease and Isolated Complex I Deficiency. Frontiers in Genetics, 2020, 11, 24. | 2.3 | 14 |
| 47 | Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589. | 6.9 | 45 |
| 48 | The feasibility of muscle mitochondrial respiratory chain phenotyping across the cognitive spectrum in Parkinson's disease. Experimental Gerontology, 2020, 138, 110997. | 2.8 | 4 |
| 49 | Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial tRNA <sup>Gly</sup> ( MT-TG ) variant. Neurology: Genetics, 2020, 6, e413. | 1.9 | 2 |
| 50 | Progressive external ophthalmoplegia due to a recurrent de novo m.15990C>T MT-TP (mt-tRNAPro) gene variant. Neuromuscular Disorders, 2020, 30, 346-350. | 0.6 | 4 |
| 51 | Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279. | 6.2 | 33 |
| 52 | Mitochondrial DNA mutations induce mitochondrial biogenesis and increase the tumorigenic potential of Hodgkin and Reedâ€“Sternberg cells. Carcinogenesis, 2020, 41, 1735-1745. | 2.8 | 10 |
| 53 | Albinism and a mitochondrial DNA deletion. Ophthalmic Genetics, 2020, 41, 295-298. | 1.2 | 1 |
| 54 | Biâ€allelic pathogenic variants in NDUFC2 cause earlyâ€onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619. | 6.9 | 17 |

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|---|---|---|---|
| 55 | Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268. | 2.9 | 19 |
| 56 | Dissecting the neuronal vulnerability underpinning Alpersâ€™ syndrome: a clinical and neuropathological study. Brain Pathology, 2019, 29, 97-113. | 4.1 | 20 |
| 57 | A novel mitochondrial m.4414T>C MT-TM gene variant causing progressive external ophthalmoplegia and myopathy. Neuromuscular Disorders, 2019, 29, 693-697. | 0.6 | 2 |
| 58 | Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776. | 2.9 | 19 |
| 59 | Resolving complexity in mitochondrial disease: Towards precision medicine. Molecular Genetics and Metabolism, 2019, 128, 19-29. | 1.1 | 25 |
| 60 | Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. Cell Reports, 2019, 26, 996-1009.e4. | 6.4 | 116 |
| 61 | A Novel Pathogenic Variant in MT-CO2 Causes an Isolated Mitochondrial Complex IV Deficiency and Late-Onset Cerebellar Ataxia. Journal of Clinical Medicine, 2019, 8, 789. | 2.4 | 11 |
| 62 | Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443. | 14.5 | 16 |
| 63 | Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748. | 2.5 | 31 |
| 64 | Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2057-2066. | 3.6 | 19 |
| 65 | A novel pathogenic m.4412G>A MT-TM mitochondrial DNA variant associated with childhood-onset seizures, myopathy and bilateral basal ganglia changes. Mitochondrion, 2019, 47, 18-23. | 3.4 | 4 |
| 66 | Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524. | 3.7 | 17 |
| 67 | Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108. | 3.3 | 12 |
| 68 | ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. PLoS Genetics, 2019, 15, e1007605. | 3.5 | 25 |
| 69 | Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. Nature Communications, 2019, 10, 759. | 12.8 | 34 |
| 70 | NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58. | 7.6 | 51 |
| 71 | Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219. | 2.8 | 26 |
| 72 | MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93. | 6.1 | 47 |

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|---|---|---|---|
| 73 | Defective mitochondrial protease LonP1 can cause classical mitochondrial disease. Human Molecular Genetics, 2018, 27, 1743-1753. | 2.9 | 46 |
| 74 | Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235. | 2.4 | 31 |
| 75 | Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504. | 6.2 | 59 |
| 76 | A novel histochemistry assay to assess and quantify focal cytochrome c oxidase deficiency. Journal of Pathology, 2018, 245, 311-323. | 4.5 | 17 |
| 77 | Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127. | 1.2 | 36 |
| 78 | Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. Scientific Reports, 2018, 8, 1799. | 3.3 | 30 |
| 79 | SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265. | 2.4 | 50 |
| 80 | Disclosing the functional changes of two genetic alterations in a patient with Chronic Progressive External Ophthalmoplegia: Report of the novel mtDNA m.7486G>A variant. Neuromuscular Disorders, 2018, 28, 350-360. | 0.6 | 10 |
| 81 | Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130. | 5.3 | 42 |
| 82 | Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6. | 9.7 | 102 |
| 83 | Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578. | 2.5 | 22 |
| 84 | Loss-of-function mutations inISCA2disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. Human Mutation, 2018, 39, 537-549. | 2.5 | 21 |
| 85 | Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345. | 3.7 | 102 |
| 86 | Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73. | 1.1 | 25 |
| 87 | Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521. | 3.2 | 73 |
| 88 | NEW OBSERVATIONS REGARDING THE RETINOPATHY OF GENETICALLY CONFIRMED KEARNSâ€“SAYRE SYNDROME. Retinal Cases and Brief Reports, 2018, 12, 349-358. | 0.6 | 8 |
| 89 | A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 Câ€methyltransferase deficiency. Human Mutation, 2018, 39, 69-79. | 2.5 | 43 |
| 90 | Further delineation of the phenotypic spectrum ofÂ ISCA2 defect: A report of ten new cases. European Journal of Paediatric Neurology, 2018, 22, 46-55. | 1.6 | 21 |

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|---|---|---|---|
| 91 | Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105. | 9.0 | 26 |
| 92 | The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42. | 1.1 | 24 |
| 93 | Expanding the clinical phenotype of IARS2-related mitochondrial disease. BMC Medical Genetics, 2018, 19, 196. | 2.1 | 16 |
| 94 | A Wars2 Mutant Mouse Model Displays OXPHOS Deficiencies and Activation of Tissue-Specific Stress Response Pathways. Cell Reports, 2018, 25, 3315-3328.e6. | 6.4 | 35 |
| 95 | Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601. | 6.2 | 41 |
| 96 | Sideroblastic anemia with myopathy secondary to novel, pathogenic missense variants in the YARS2 gene. Haematologica, 2018, 103, e564-e566. | 3.5 | 5 |
| 97 | Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. Human Molecular Genetics, 2018, 27, 4135-4144. | 2.9 | 30 |
| 98 |  <scp>OXA</scp> 1L  mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, . | 6.9 | 54 |
| 99 | Skeletal muscle mitochondrial oxidative phosphorylation function in idiopathic pulmonary arterial hypertension: in vivo and in vitro study. Pulmonary Circulation, 2018, 8, 1-5. | 1.7 | 10 |
| 100 | Expanding the phenotype of de novo SLC25A4 -linked mitochondrial disease to include mild myopathy. Neurology: Genetics, 2018, 4, e256. | 1.9 | 20 |
| 101 | Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231. | 6.2 | 65 |
| 102 | Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114. | 6.2 | 34 |
| 103 | Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. Annals of Neurology, 2018, 84, 289-301. | 5.3 | 47 |
| 104 | mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, . | 6.9 | 199 |
| 105 | Confirming TDP2 mutation in spinocerebellar ataxia autosomal recessive 23 (SCAR23). Neurology: Genetics, 2018, 4, e262. | 1.9 | 27 |
| 106 | Inherited pathogenic mitochondrial DNA mutations and gastrointestinal stem cell populations. Journal of Pathology, 2018, 246, 427-432. | 4.5 | 13 |
| 107 | Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in NDUFB11. Journal of Physical Education and Sports Management, 2017, 3, a001271. | 1.2 | 19 |
| 108 | MR-1S Interacts with PET100 and PET117 in Module-Based Assembly of Human Cytochrome c Oxidase. Cell Reports, 2017, 18, 1727-1738. | 6.4 | 86 |

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|---|---|---|---|
| 109 | Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686. | 9.0 | 41 |
| 110 | Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275. | 6.2 | 217 |
| 111 | Camptocormia and shuffling gait due to a novel MT-TV mutation: Diagnostic pitfalls. Neurology: Genetics, 2017, 3, e147. | 1.9 | 3 |
| 112 | Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824. | 12.8 | 432 |
| 113 | Clinically proven mtDNA mutations are not common in those with chronic fatigue syndrome. BMC Medical Genetics, 2017, 18, 29. | 2.1 | 15 |
| 114 | De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83. | 3.2 | 54 |
| 115 | Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159. | 6.2 | 63 |
| 116 | De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187. | 1.9 | 11 |
| 117 | Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235. | 1.4 | 10 |
| 118 | Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538. | 6.2 | 58 |
| 119 | Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164. | 2.8 | 18 |
| 120 | Pigmentary retinopathy, rodâ€“cone dysfunction and sensorineural deafness associated with a rare mitochondrial tRNALys(m.8340G>A) gene variant. British Journal of Ophthalmology, 2017, 101, 1298-1302. | 3.9 | 8 |
| 121 | Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330. | 5.3 | 65 |
| 122 | Multipotent Basal Stem Cells, Maintained in Localized Proximal Niches, Support Directed Long-Ranging Epithelial Flows in Human Prostates. Cell Reports, 2017, 20, 1609-1622. | 6.4 | 64 |
| 123 | Novel POLG variants associated with late-onset de novo status epilepticus and progressive ataxia. Neurology: Genetics, 2017, 3, e181. | 1.9 | 2 |
| 124 | Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015. | 5.3 | 63 |
| 125 | Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. Scientific Reports, 2017, 7, 15676. | 3.3 | 20 |
| 126 | The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250. | 4.5 | 329 |

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|---|---|---|---|
| 127 | Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130. | 3.6 | 23 |
| 128 | Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202. | 1.9 | 1 |
| 129 | Nucleotide pools dictate the identity and frequency of ribonucleotide incorporation in mitochondrial DNA. PLoS Genetics, 2017, 13, e1006628. | 3.5 | 55 |
| 130 | Compound heterozygous RMND1 gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. BMC Research Notes, 2016, 9, 325. | 1.4 | 15 |
| 131 | Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227. | 6.2 | 57 |
| 132 | Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113. | 1.9 | 12 |
| 133 | The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775. | 3.2 | 35 |
| 134 | P35â€…Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: Phenotypes linked by truncating variants inNDUFB11. Heart, 2016, 102, A18.2-A18. | 2.9 | 0 |
| 135 | The frequency of the m.1555Aâ€‰>â€‰G (MTRNR1) variant in UK patients with suspected mitochondrial deafness. Hearing, Balance and Communication, 2016, 14, 101-102. | 0.4 | 14 |
| 136 | Cystic Leukoencephalopathy due to NDUFV1 mutationâ€”A Report of the Phenotype and Its Rare Co-occurrence with Primary Hyperoxaluria. Journal of Pediatric Neurology, 2016, 14, 126-132. | 0.2 | 0 |
| 137 | A leaky splicing mutation in NFU1 is associated with a particular biochemical phenotype. Consequences for the diagnosis. Mitochondrion, 2016, 26, 72-80. | 3.4 | 19 |
| 138 | Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59. | 1.9 | 86 |
| 139 | Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131. | 3.2 | 91 |
| 140 | Clinical features of the pathogenic m.5540G>A mitochondrial transfer RNA tryptophan gene mutation. Neuromuscular Disorders, 2016, 26, 702-705. | 0.6 | 6 |
| 141 | Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24. | 4.1 | 22 |
| 142 | Impaired mitochondrial biogenesis is a common feature to myocardial hypertrophy and end-stage ischemic heart failure. Cardiovascular Pathology, 2016, 25, 103-112. | 1.6 | 77 |
| 143 | Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. Nucleic Acids Research, 2016, 44, 5313-5329. | 14.5 | 37 |
| 144 | Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668. | 9.0 | 69 |

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|---|---|---|---|
| 145 | Resveratrol attenuates oxidative stress in mitochondrial Complex I deficiency: Involvement of SIRT3. Free Radical Biology and Medicine, 2016, 96, 190-198. | 2.9 | 47 |
| 146 | The m.13051G>A mitochondrial DNA mutation results in variable neurology and activated mitophagy. Neurology, 2016, 86, 1921-1923. | 1.1 | 35 |
| 147 | Dysferlin mutations and mitochondrial dysfunction. Neuromuscular Disorders, 2016, 26, 782-788. | 0.6 | 28 |
| 148 | Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876. | 6.2 | 93 |
| 149 | Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632. | 9.7 | 241 |
| 150 | Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82. | 1.9 | 24 |
| 151 | Mitochondrial dysfunction in myofibrillar myopathy. Neuromuscular Disorders, 2016, 26, 691-701. | 0.6 | 32 |
| 152 | Investigating complex <scp>I</scp> deficiency in <scp>P</scp>urkinje cells and synapses in patients with mitochondrial disease. Neuropathology and Applied Neurobiology, 2016, 42, 477-492. | 3.2 | 23 |
| 153 | Lethal Neonatal LTBL Associated with Biallelic EARS2 Variants: Case Report and Review of the Reported Neuroradiological Features. JIMD Reports, 2016, 33, 61-68. | 1.5 | 23 |
| 154 | Epilepsy due to mutations in the mitochondrial polymerase gamma (<scp>POLG</scp>) gene: A clinical and molecular genetic review. Epilepsia, 2016, 57, 1531-1545. | 5.1 | 58 |
| 155 | Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682. | 6.2 | 48 |
| 156 | Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692. | 5.3 | 40 |
| 157 | Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. Nature Communications, 2016, 7, 12317. | 12.8 | 106 |
| 158 | The swinging pendulum of biomarkers in mitochondrial disease. Neurology, 2016, 87, 2286-2287. | 1.1 | 5 |
| 159 | A recurrent mitochondrial p.Trp22ArgNDUFB3variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. Journal of Medical Genetics, 2016, 53, 634-641. | 3.2 | 31 |
| 160 | Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000. | 6.2 | 89 |
| 161 | Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33. | 7.6 | 15 |
| 162 | Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16. | 3.6 | 92 |

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|---|---|---|---|
| 163 | Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041. | 2.9 | 53 |
| 164 | Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. European Heart Journal, 2016, 37, 2552-2559. | 2.2 | 53 |
| 165 | MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779. | 3.5 | 67 |
| 166 | Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957. | 5.3 | 62 |
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